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Glucose-6-Phosphate Dehydrogenase Deficiency: A Single-Center Experience

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In their interesting study, Kılıç et al[1] studied the demographic, clinical, and laboratory spectra of 65 patients with glucose-6-phosphate dehydrogenase (G6PD) deficiency admitted to the pediatric hematology and oncology clinic in Turkey between 2007 and 2019. They found that the disease was more common in boys (90.7%) than in girls (9.2%). The mean value of the G6PD enzyme on admission was 1.9 ± 1.4 IU/g of hemoglobin (Hb), and a positive family history of the disease was found in 40% of the patients. Prolonged jaundice was the most common presentation, and the most common physical finding was jaundice while splenomegaly was not detected. Cholecystectomy, splenectomy, and regular erythrocyte transfusion were not reported during follow-up. Kılıç et al[1] mentioned 2 study limitations. There was a partial lack of patient data in the electronic recording systems, and patient files and genetic analysis of the G6PD variant could not be accomplished. I presume that the following limitation is relevant. It is explicit that apart from the suggestive clinical presentation, confirming the diagnosis of the G6PD disease requires the laboratory demonstration of a low erythrocyte G6PD enzyme level. Since the G6PD enzyme level is ethnicity and population-specific,[2,3] many populations have constructed their reference values of the G6PD enzyme. Interestingly, Turkey is a pioneer country that has set its standard of G6PD enzyme levels.[4] The values of the G6PD enzyme in different age groups of people in Turkey have been determined. The normal mean values of G6PD in healthy females were 9.16 ± 3.78 IU/g Hb and 8.94 ± 8.65 IU/g Hb in males.[4] In the study methodology, Kılıç et al[1] stated that the disease was diagnosed by the suspected clinical picture and low G6PD enzyme levels. The laboratory normal range for the G6PD enzyme level employed in the study was 7-20.5 IU/g of Hb. However, Kılıç et al[1] did not address the reference for the utilized laboratory normal values of the G6PD enzyme. The noticeable difference in the normal G6PD values between Turkish standard[4] and that employed in the study by Kılıç et al[1] might question the actual number of confirmed G6PD patients recruited in the study. Hence, it might cast further suspicions on the accurateness of the study results.

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